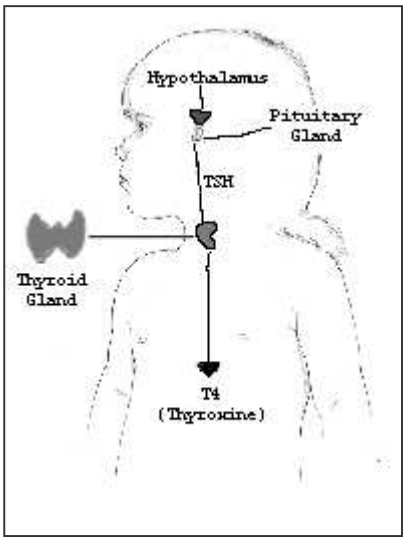
Congenital Hypothyroidism (CH)
General Overview

Q. What is CH?

A. It is a condition caused by insufficient production of the thyroid hormone, thyroxine (T4), as a result of malformation or malfunction of the thyroid gland. Treatment is required for CH in order to prevent problems with growth and development.

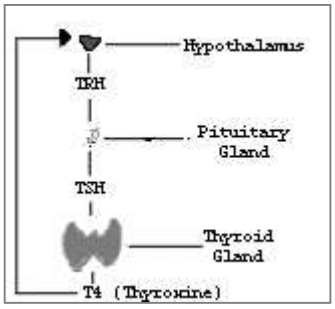


Q. Where is the thyroid gland and what does it do?

A. The thyroid is a small, butterfly-shaped gland located in the front of the neck. It takes up iodine from the food we eat and uses it to make thyroid hormone, also called thyroxine or "T4". Thyroid hormone plays a vital role in normal growth and development in children. If the thyroid gland has not developed properly, it will not produce enough T4 for normal body growth and brain development.

Q. How does the thyroid gland normally produce T4?

A. The process of producing T4 begins in the brain. It is controlled by the pituitary, or "master gland". The pituitary gland is a pea-sized gland that is found at the base of the brain just below the hypothalamus. It receives signals from the hypothalamus which stimulates the release of hormones, which in turn affect many of the body's functions. One of the hormones that the pituitary gland produces is called thyroid stimulating hormone (TSH). TSH encourages growth of the thyroid gland as well as stimulating it to release T4. The release of TSH from the pituitary is triggered by another hormone called thyrotrophin-releasing hormone (TRH) which is released from the hypothalamus. These glands and hormones control the levels of thyroxine in the body as follows:



If levels of thyroxine in the body are low, this is detected by the hypothalamus and TRH is released. The TRH stimulates the pituitary gland to produce more TSH and this triggers the thyroid to increase the amount of thyroxine released.

If levels of thyroxine are high, production of TRH and TSH are stopped and so the thyroid gland stops releasing thyroxine until it is again required to do so.

Q. What causes the thyroid gland not to function properly in children with CH?

A. The most common cause for the thyroid not to function properly is improper growth of the gland. During early pregnancy the thyroid gland in the developing baby begins to form at the base of the brain and then moves to its place in the lower neck where it finishes growing. Sometimes that process is interrupted (for reasons not yet known), leaving only a small piece of thyroid which may be abnormal in location. Some babies will have no thyroid gland at all. These forms of hypothyroidism are permanent conditions which the child will not outgrow. Another, less common cause of thyroid gland malfunction is due to lack of stimulation by the pituitary gland.

Q. What are the effects of having CH?

A. Untreated CH can lead to severe, permanent, neurological and developmental damage.

Q. Is there only one form of CH?

A. There are two main categories of CH, developmental and enzymatic. Developmental CH occurs when the thyroid gland does not reach its proper position (ectopic) or when the gland does not fully develop or fails to develop at all (agenesis). Enzymatic CH occurs when the thyroid gland is present, and may even be enlarged, but it fails to produce thyroxine. This form of CH is much more likely to be inherited and is called a dyshormonogenesis, which means there is a block that prevents the hormones being produced.

Q. What are the chances that a child will be born with CH?

A. About one in every 5,000 babies is born with CH.

Q. What is the treatment for CH?

A. The treatment for CH is daily replacement of missing thyroid hormone with synthetic thyroid hormone tablets, synthroid. The synthetic thyroid hormone acts exactly like the hormone produced by the thyroid gland. When given at the proper dosage, there are usually no side effects and the child will have normal development.

Washington State Department of Health
Office of Newborn Screening
1610 NE 150th Street, K17-9
Shoreline, WA 98155-0729
(206) 361-2902 FAX (206) 361-4996